Orofacial function of persons having RubinsteinTaybi syndrome

Report from observation charts

The survey comprises 33 observation charts.

Estimated occurrence: 1-2:100 000 live births.

Etiology: Usually damage to the short arm of chromosome 16 (16p 13.3) but damage on chromosome 22 (22q13) has also been detected. The genes affected (CBP and EP300) are crucial for the development of different organ systems during fetal life. Diagnosis made on the basis of symptoms, and in about one half of individuals this can be confirmed with DNA analysis.

General symptoms: Characteristic for the diagnosis is short stature even from birth, broad thumbs and big toes turned outwards and a prominent nose. Vertebral deformations as well as anomalies of the sternum and ribs are often visible on X-ray. Deformation of internal organs such as cardiac and urinary tract defects is common and so are hyper mobility of joints, funnel chest, scoliosis and lordosis. Many children have vision problems and hearing impairments as a result of infections, attributable to an increased sensitivity to infection. Intellectual disability and delayed motor development are included in the clinical picture and many have difficulties with concentration and hyperactivity.

Orofacial/odontological symptoms: There are characteristic facial features that become more distinct with age. Small jaws and a high, arched palate are frequent, as are crowded teeth. Some teeth may curve outwards, a condition known as "talon cusp". Many of these children have eating difficulties and delayed speech and language development. Drooling may occur.

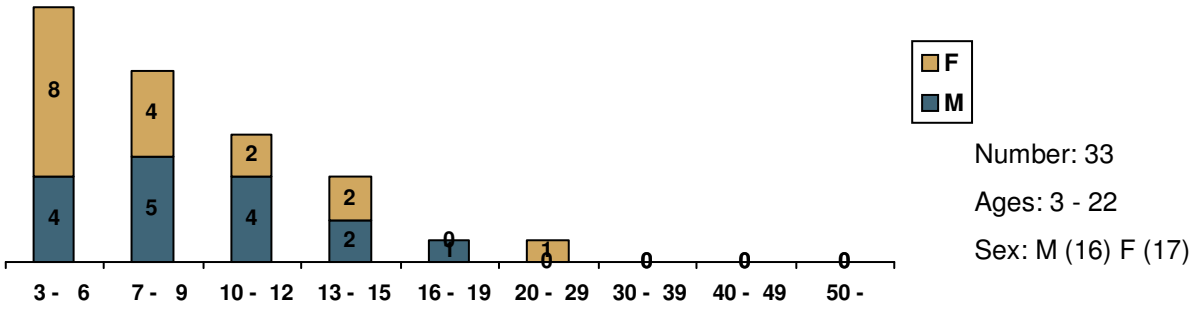
Orofacial/ odontological treatment:

- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- Orofacial therapy and oral motor skills training could be relevant in cases of difficulties with eating, speech or drooling.
- Speech, language and communication training are often justified.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.
- Caution should be shown in cases of intubation for anaesthetized dental care, owing to the possibility of instability in the cervical vertebral column.
- When treating medically compromised patients always contact their doctors for medical advice.

Sources:

The rare disease database of the Swedish National Board of Health and Welfare.
The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.
The Documentation from the Ågrenska Center.

Age distribution



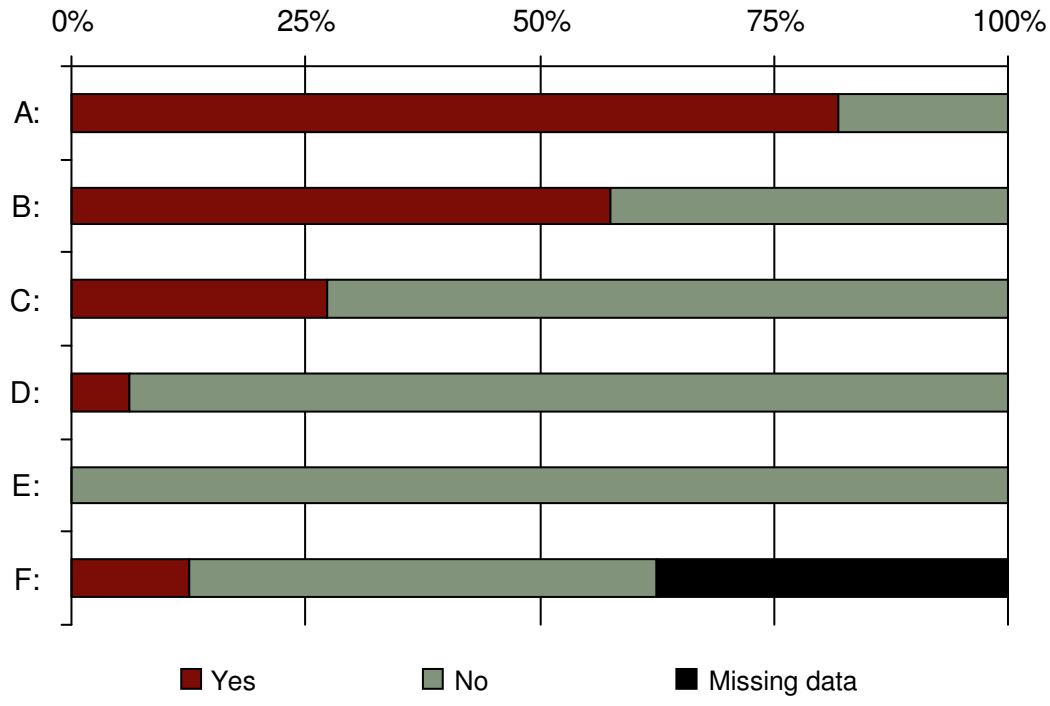
Number: 33

Ages: 3 - 22

Sex: M (16) F (17)

Overview

		Yes	No	Missing data	N
A:	Incomprehensible speech/No speech	27	6	0	33
B:	Eating and drinking difficulties[1]	19	14	0	33
C:	Profuse drooling, on clothes[1]	9	24	0	33
D:	Breathing difficulties[1] [2]	1	15	0	16
E:	Grinding every day[1] [2]	0	16	0	16
F:	Severe malocclusions[2]	2	8	6	16



Note that the diagram is based upon less than 100 individuals.

1: Compiled using questionnaire

2: This variable was introduced in version 2 (2008) of the Observation chart.

Oral health

Oral health index (indices)[1]

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	3	0	0	0	0	0	0	13	16
GI	Gingivitis	3	0	0	0	0	0	0	13	16
Plaq	Coating	2	0	0	0	0	0	1	13	16
Toot	Tooth wear	1	1	0	0				14	16

C Calculus index is based on the presence of visible calculus on the buccal surface of 6 index teeth. 0 indicates that there is no calculus at all, 6 indicates calculus on all index teeth.

GI Gingivitis index is based on the presence of visible gingivitis on the buccal surface of 6 index teeth. 0 indicates that there is no bleeding, 6 indicates bleeding on all index teeth.

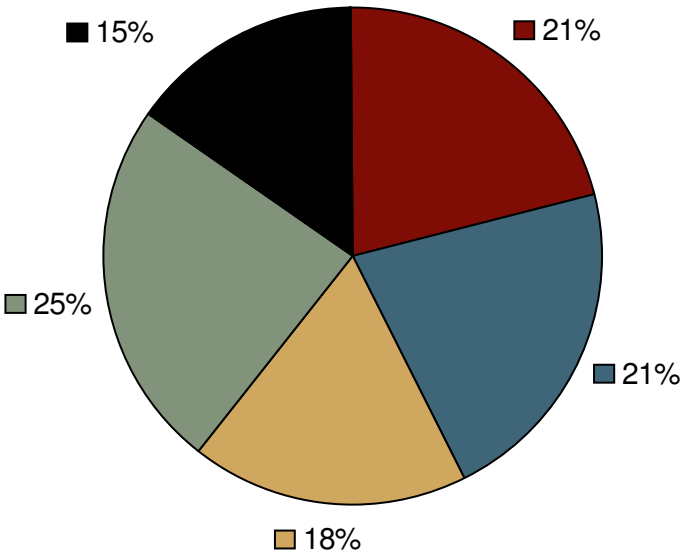
PI Plaque index is based on the presence of visible plaque on the buccal surface of 6 index teeth. 0 indicates that there is no plaque, 6 indicates plaque on all index teeth.

To Tooth wear index is a weighted summary of the degree of tooth wear on 6 different segments. Tooth wear is only evaluated in the permanent dentition, not in the primary teeth. The final index score is based on the degree of tooth wear found in most segments.

0: No tooth wear or minor wear of enamel in either of the segments
1: Marked tooth wear of the enamel, possibly exceeding into dentin
2: tooth wear in the dentine reaching up to 1/3 of the tooth crown
3: Tooth wear in the dentine reaching up to more than 1/3 of the tooth crown. If 3 is given in any segment then SI is 3.

1: Oral health index (indices) was (were) introduced in the observations in 2008

Acceptance of dental examination



	Number
None	7
Negative	7
Reluctant	6
Positive	8
Missing data	5
Sum:	**33**

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft[1]				
Examined	8	6		
Number of individuals with deft=0	6	6		
Mean	0,5	0,0		
Standard deviation	1,0	0,0		
Missing data	4	9		
DMFT[2]				
Examined		7	1	1
Number of individuals with DMFT=0		7	1	1
Standard deviation		0,0	0,0	0,0
Mean		0,0	0,0	0,0
Missing data		8	4	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	9
Post normal	6
Pre normal	5
Missing data	13
	Sum: 33

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	1
31 - 40	2
41 - 50	0
51 -	1
Missing data	17
	Sum: 21

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	1
31 - 40	4
41 - 50	2
51 -	0
Missing data	5
	Sum: 12

Profile[1]

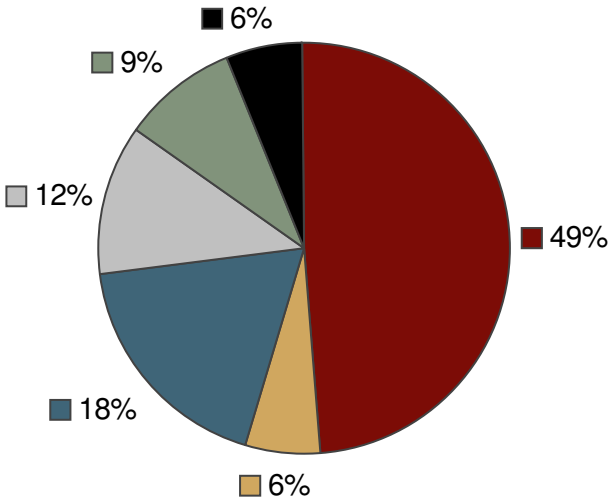
	Number
Normal	9
Convex	4
Concave	0
Missing data	3
	Sum: 16

Mandibular plane[1]

	Number
Normal	12
Increased	1
Reduced	0
Missing data	3
	Sum: 16

1: This variable was introduced in version 2 (2008) of the Observation chart.

Speech difficulty



	Number
No speech	16
Very incomprehensible	2
Incomprehensible speech	6
Slightly indistinct speech	4
No problems	3
Missing data	2

Sum: 33

Clinical findings	Yes-answers Total N=33 (%)	Boys/Men N=16 (%)	Girls/Women N=17 (%)	Missing data
Open mouth at rest	**23** (70)	12 (75)	11 (65)	0
Narrow palate	**20** (71)	10 (77)	10 (67)	5
Low muscle tone in lips	**17** (52)	12 (75)	5 (29)	0
High palate	**16** (57)	8 (62)	8 (53)	5
Impaired tongue motility	**12** (41)	6 (43)	6 (40)	4
Over crowding	**8** (40)	3 (38)	5 (42)	13
Low muscle tone in tongue	**6** (25)	4 (44)	2 (13)	9
Frontal open bite	**5** (22)	1 (11)	4 (29)	10
Reduced stability in neck	**4** (12)	1 (6)	3 (18)	0
M mentalis overactive	**4** (13)	2 (13)	2 (12)	1